



RPS17 gene

ribosomal protein S17

Normal Function

The *RPS17* gene provides instructions for making one of approximately 80 different ribosomal proteins, which are components of cellular structures called ribosomes. Ribosomes process the cell's genetic instructions to create proteins.

Each ribosome is made up of two parts (subunits) called the large and small subunits. The protein produced from the *RPS17* gene is among those found in the small subunit.

The specific functions of the RPS17 protein and the other ribosomal proteins within these subunits are unclear. Some ribosomal proteins are involved in the assembly or stability of ribosomes. Others help carry out the ribosome's main function of building new proteins. Studies suggest that some ribosomal proteins may have other functions, such as participating in chemical signaling pathways within the cell, regulating cell division, and controlling the self-destruction of cells (apoptosis).

Health Conditions Related to Genetic Changes

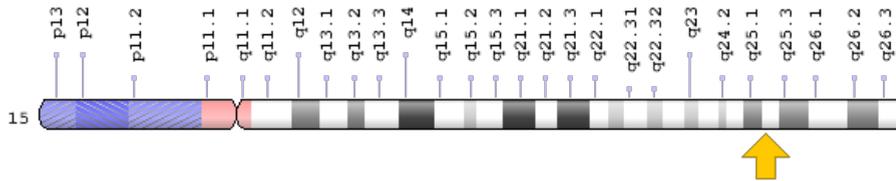
Diamond-Blackfan anemia

At least three *RPS17* gene mutations have been identified in individuals with Diamond-Blackfan anemia. These mutations are believed to affect the stability or function of the RPS17 protein. Studies indicate that a shortage of functioning ribosomal proteins may increase the self-destruction of blood-forming cells in the bone marrow, resulting in a low number of red blood cells (anemia). Abnormal regulation of cell division or inappropriate triggering of apoptosis may contribute to the other health problems and unusual physical features that affect some people with Diamond-Blackfan anemia.

Chromosomal Location

Cytogenetic Location: 15q25.2, which is the long (q) arm of chromosome 15 at position 25.2

Molecular Location: base pairs 82,536,750 to 82,540,544 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 40S ribosomal protein S17
- DBA4
- MGC72007
- RPS17L1
- RPS17L2
- RS17_HUMAN
- S17

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The RNA message is decoded on ribosomes
<https://www.ncbi.nlm.nih.gov/books/NBK26829/#A1071>

GeneReviews

- Diamond-Blackfan Anemia
<https://www.ncbi.nlm.nih.gov/books/NBK7047>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RPS17%5BTIAB%5D%29+OR+%28ribosomal+protein+S17%5BTIAB%5D%29%29+OR+%28%28DBA4%5BTIAB%5D%29+OR+%2840S+ribosomal+protein+S17%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- RIBOSOMAL PROTEIN S17
<http://omim.org/entry/180472>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RPS17%5Bgene%5D>
- Diamond-Blackfan Anemia Mutation Database
http://www.dbagenes.unito.it/home.php?select_db=RPS17
- HGNC Gene Family: S ribosomal proteins
<http://www.genenames.org/cgi-bin/genefamilies/set/728>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10397
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6218>
- UniProt
<http://www.uniprot.org/uniprot/P08708>

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